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Carriers of inversions produce from 0 to 54.3 per cent abnormal sperm. Carriers of Robertsonian translocations produce from 3.4 to 36.0 per cent abnormal sperm, and carriers of reciprocal translocations produce from

47.5 to 81.0 per cent abnormal spermatozoa. However, carriers of translocations usually produce more abnormal embryos than expected from these figures. This may be partly related to interchromosomal effects induced by some structural reorganizations. Males with oligoasthenozoospermia, low motility and/or high FSH concentrations show frequent synaptic anomalies, resulting in the production of aneuploid and diploid sperm. Testicular sperm show extremely high rates of chromosomal abnormalities. The risk of recurrent abortion is increased by the presence of chromosome abnormalities in sperm.

L5 ANSWER 2 OF 17 MEDLINE on STN ACCESSION NUMBER: 2001530620 MEDLINE DOCUMENT NUMBER: PubMed ID: 11576728

TITLE: Studies on sperm chromosomes in patients with severe

male factor infertility undergoing

assisted reproductive technology treatment.

AUTHOR: Levron J; Aviram-Goldring A; Madgar I; Raviv G; Barkai G;

Dor J

CORPORATE SOURCE: Department of Obstetrics and Gynecology, The Chaim Sheba

Medical Center, Tel Hashomer 52621, Israel.

SOURCE: Molecular and cellular endocrinology, (2001 Oct 22)

Vol. 183 Suppl 1, pp. S23-8.

Journal code: 7500844. ISSN: 0303-7207.

PUB. COUNTRY: Ireland

DOCUMENT TYPE: (COMPARATIVE STUDY)

Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 200308

ENTRY DATE: Entered STN: 1 Oct 2001

Last Updated on STN: 11 Dec 2002 Entered Medline: 29 Aug 2003

AΒ The aim of the study was to determine the rate of chromosome abnormalities in testicular sperm after intracytoplasmic sperm injection due to severe male factor infertility. The study groups included patient with non-obstructive azoospermia (n=9), obstructive azoospermia (n=10), Klinefelter's syndrome (n=5) and normal controls (n=6, groups I-VI, respectively). The mean serum levels of FSH 17.5+/-8.2(P<0.05), 3.5+/-2.6, 29.8+/-13.0 (P<0.05) and 3.1+/-0.4 mIU/ml, respectively. The rates of chromosome abnormalities were 19.6% (P<0.001), 8.2% (P<0.001), 6.3 and 1.6%, respectively. Chromosomes X and Y were significantly more involved in the aneuploidy than chromosome 18 in groups I and II. The present findings demonstrate a linkage between gonadal failure (high serum FSH levels) and sperm chromosome abnormalities. Our findings may explain the increased incidence of perinatal sex chromosome abnormalities found in severe male factor patients. Patients with non-mosaic Klinefelter's syndrome have comparable risk for sex chromosomes aneuploidy as the rest of the patients with azoospermia. Therefore, genetic screening during pregnancy or before embryo replacement should be carefully considered in severe male factor patient following in vitro fertilization (IVF).

L5 ANSWER 3 OF 17 MEDLINE on STN ACCESSION NUMBER: 2001490169 MEDLINE DOCUMENT NUMBER: PubMed ID: 11532468

TITLE: Sperm chromosome abnormalities in men with severe

male factor infertility who are

undergoing in vitro fertilization with intracytoplasmic

sperm injection.

AUTHOR: Levron J; Aviram-Goldring A; Madgar I; Raviv G; Barkai G;

Dor J

CORPORATE SOURCE: IVF Unit, Division of Obstetrics and Gynecology, The Chaim

Sheba Medical Center, Tel-Hashomer, Israel..

jlevron@netvision.net.il

SOURCE: Fertility and sterility, (2001 Sep) Vol. 76, No.

3, pp. 479-84.

Journal code: 0372772. ISSN: 0015-0282.

PUB. COUNTRY: United States
DOCUMENT TYPE: (COMPARATIVE STUDY)

Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 200110

ENTRY DATE: Entered STN: 5 Sep 2001

Last Updated on STN: 15 Oct 2001 Entered Medline: 11 Oct 2001

AΒ OBJECTIVE: To investigate the potential paternal contribution to the risk of fetal chromosomal anomalies after intracytoplasmic sperm injection (ICSI). DESIGN: Spermatozoa isolated from testicular tissue and ejaculated specimens of consenting patients undergoing testicular biopsy and ICSI were analyzed for chromosomes X, Y, and 18 by FISH. SETTING: Assisted reproductive technology program. PATIENT(S): Consenting patients undergoing testicular biopsy and ICSI, severe oligozoospermic patients, and normal fertile donors. INTERVENTION(S): None. MAIN OUTCOME MEASURE(S): The rate of chromosome abnormalities in testicular sperm with regard to the type of azoospermia and ejaculated sperm compared to healthy men. RESULT(S): The mean serum levels of FSH in the groups with nonobstructive azoospermia (n = 9), obstructive azoospermia (n = 10), severe oligozoospermia (n = 9), and the normal donors (n = 6) were 17.5+/- 8.2 (P<.05), 3.5 +/- 2.6, 14.6 +/- 3.5 (P<.05), and 3.1 +/- 0.4 IU/mL, respectively. The corresponding rates of sperm chromosome abnormalities among these groups were 19.6% (P<.001), 8.2% (P<.001), 13.0% (P<.001), and 1.6%, respectively. The corresponding rates of disomy among these groups were 7.8% (12 of 153 spermatozoa), 4.9% (18 of 367), 6.2% (109 of 1,751), and 1% (5 of 500 spermatozoa), respectively. Errors in chromosomes X and Y were significantly more common than in chromosome 18. CONCLUSION(S): The present findings demonstrate a linkage between gonadal failure (high serum FSH levels) and the occurrence of sperm chromosome aneuploidies. Our findings may explain the increased incidence of sex chromosome abnormalities found after IVF in the severe male factor patient population. Genetic screening during pregnancy or before embryo replacement should be considered carefully.

L5 ANSWER 4 OF 17 MEDLINE on STN ACCESSION NUMBER: 2000247304 MEDLINE DOCUMENT NUMBER: PubMed ID: 10783364

TITLE: Chromosome analysis of spermatozoa extracted from testes of

men with non-obstructive azoospermia.

AUTHOR: Martin R H; Greene C; Rademaker A; Barclay L; Ko E; Chernos

J

CORPORATE SOURCE: Department of Medical Genetics, Faculty of Medicine,

University of Calgary, Alberta, Canada.

SOURCE: Human reproduction (Oxford, England), (2000 May)

Vol. 15, No. 5, pp. 1121-4.

Journal code: 8701199. ISSN: 0268-1161.

PUB. COUNTRY: ENGLAND: United Kingdom

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE) (RESEARCH SUPPORT, NON-U.S. GOV'T)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 200007

ENTRY DATE: Entered STN: 28 Jul 2000

Last Updated on STN: 13 Aug 2001 Entered Medline: 20 Jul 2000

AB Infertile men with azoospermia now have the possibility of fathering children by testicular sperm extraction combined with intracytoplasmic sperm injection. However, there are concerns about the risk of chromosomal abnormalities in their spermatozoa. We have studied aneuploidy frequencies for chromosomes 13, 21, X and Y by multicolour fluorescence in-situ hybridization (FISH) in testicular spermatozoa extracted from three men with non-obstructive azoospermia. The men were 34-37 years of age and had normal folliclestimulating hormone (FSH) concentrations and normal 46,XY somatic karyotypes. A total of 3324 spermatozoa was analysed. The infertile patients had an elevated frequency of disomy for chromosomes 13, 21, XY disomy compared to controls but none of these reached statistical significance. Also there was no significant difference in the sex ratio or the frequency of diploidy in azoospermic patients compared to normal control donors. This first report on chromosomal aneuploidy in spermatozoa extracted from testes $% \left(1\right) =\left(1\right) \left(1\right) +\left(1\right) \left(1\right) \left(1\right) +\left(1\right) \left(1\right) \left($ of patients with non-obstructive azoospermia suggests that some azoospermic men do not have a substantially increased risk of chromosomally abnormal spermatozoa.

L5 ANSWER 5 OF 17 MEDLINE on STN ACCESSION NUMBER: 2000174998 MEDLINE DOCUMENT NUMBER: PubMed ID: 10711834

TITLE: Human male infertility: chromosome

anomalies, meiotic disorders, abnormal spermatozoa and

recurrent abortion.

AUTHOR: Egozcue S; Blanco J; Vendrell J M; Garcia F; Veiga A; Aran

B; Barri P N; Vidal F; Egozcue J

CORPORATE SOURCE: Departament de Biologia Cellular, Universitat Autonoma de

Barcelona, Bellaterra, Spain.

SOURCE: Human reproduction update, (2000 Jan-Feb) Vol. 6,

No. 1, pp. 93-105. Ref: 146

Journal code: 9507614. ISSN: 1355-4786.

PUB. COUNTRY: ENGLAND: United Kingdom

DOCUMENT TYPE: (CASE REPORTS)

Journal; Article; (JOURNAL ARTICLE)

General Review; (REVIEW)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 200004

ENTRY DATE: Entered STN: 27 Apr 2000

Last Updated on STN: 27 Apr 2000 Entered Medline: 19 Apr 2000

Human male infertility is often related to chromosome AB abnormalities. In chromosomally normal infertile males , the rates of chromosome 21 and sex chromosome disomy in spermatozoa are increased. Higher incidences of trisomy 21 (seldom of paternal origin) and sex chromosome aneuploidy are also found. XXY and XYY patients produce increased numbers of XY, XX and YY spermatozoa, indicating an increased risk of production of XXY, XYY and XXX individuals. Since XXYs can reproduce using intracytoplasmic sperm injection (ICSI), this could explain the slight increase of sex chromosome anomalies in ICSI series. Carriers of structural reorganizations produce unbalanced spermatozoa, and risk having children with duplications and/or deficiencies. In some cases, this risk is considerably lower or higher than average. These patients also show increased diploidy, and a higher risk of producing diandric triploids. Meiotic disorders are frequent in infertile males, and increase with severe oligoasthenozoospemia (OA) and/or high follicle stimulating hormone (FSH) concentrations.

These patients produce spermatozoa with autosomal and sex chromosome disomies, and diploid spermatozoa. Their contribution to recurrent abortion depends on the production of trisomies, monosomies and of triploids. The most frequent sperm chromosome anomaly in infertile males is diploidy, originated by either meiotic mutations or by a compromised testicular environment.

L5 ANSWER 6 OF 17 MEDLINE ON STN ACCESSION NUMBER: 1998428847 MEDLINE DOCUMENT NUMBER: PubMed ID: 9757880

TITLE: Prevalence of Y chromosome microdeletions in oligospermic

and azoospermic candidates for intracytoplasmic sperm

injection.

AUTHOR: Oliva R; Margarit E; Ballesca J L; Carrio A; Sanchez A;

Mila M; Jimenez L; Alvarez-Vijande J R; Ballesta F

CORPORATE SOURCE: Genetics Service, Hospital Clinic I Provincial of

Barcelona, Faculty of Medicine, University of Barcelona,

Spain.. oliva@medicina.ub.es

SOURCE: Fertility and sterility, (1998 Sep) Vol. 70, No.

3, pp. 506-10.

Journal code: 0372772. ISSN: 0015-0282.

PUB. COUNTRY: United States
DOCUMENT TYPE: (CLINICAL TRIAL)

(CONTROLLED CLINICAL TRIAL)

Journal; Article; (JOURNAL ARTICLE) (RESEARCH SUPPORT, NON-U.S. GOV'T)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 199810

ENTRY DATE: Entered STN: 21 Oct 1998

Last Updated on STN: 21 Oct 1998

Entered Medline: 9 Oct 1998

AB OBJECTIVE: To determine the prevalence and type of Y chromosome microdeletions in 136 consecutively seen intracytoplasmic sperm injection (ICSI) candidates and in 50 consecutively seen azoospermic men attending an infertility clinic. DESIGN: Controlled clinical study. SETTING: Genetics laboratory and infertility clinic at a University hospital. PATIENT(S): One hundred eighty-six men who were seen at an infertility clinic and who were referred to a genetics counseling service for genetic assessment before ICSI. INTERVENTION(S): Collection of semen and blood samples. MAIN OUTCOME MEASURE(S): Semen analysis; serum FSH, LH, and T levels; karyotype analysis; and presence or absence of several single tagged site markers along the Y chromosome (sY274, sY238, sY276, sY84, sY102, sY143, sY153, sY254, sY269, sy202, sy158, sy160). RESULT(S): Yq chromosome microdeletions were detected in 10 (5.4%) of 186 consecutively seen ICSI candidates. The number of microdeletions was much higher in azoospermic patients (16%; 8 of 50) than in oligospermic patients (1.5%; 2 of 136). Two of the azoospermic patients with a Yq microdeletion also had sex chromosome aneuploidy mosaicism. No microdeletions were detected in 100 consecutively seen fathers who were included as controls. CONCLUSION(S): The prevalence of Yq microdeletions in the azoospermic group was much higher than in the oligospermic group and was consistent with the prevalence of Yq microdeletions detected in other series of azoospermic men in different geographic areas. All Yq microdeletions found in our patients belong to the AZFc region, indicating that microdeletions of the AZFa and AZFb regions are infrequent among oligospermic ICSI candidates or azoospermic males in our population.

L5 ANSWER 7 OF 17 MEDLINE on STN ACCESSION NUMBER: 1998401619 MEDLINE DOCUMENT NUMBER: PubMed ID: 9731432

[Contribution of chromosomal abnormalities to in vitro TITLE:

fertilization failures].

Contribucion de las anomalias cromosomicas ovocitarias en

el fracaso de la fecundacion humana in vitro.

Smith R; Walker L; Cobo A C; Vantman D AUTHOR:

CORPORATE SOURCE: Instituto de Investigaciones Materno-Infantil, Facultad de

Medicina, Universidad de Chile, Santiago, Chile.

SOURCE: Revista medica de Chile, (1998 May) Vol. 126, No.

5, pp. 511-9.

Journal code: 0404312. ISSN: 0034-9887.

PUB. COUNTRY: Chile

DOCUMENT TYPE: (ENGLISH ABSTRACT)

Journal; Article; (JOURNAL ARTICLE) (RESEARCH SUPPORT, NON-U.S. GOV'T)

LANGUAGE: Spanish

FILE SEGMENT: Priority Journals

ENTRY MONTH: 199811

ENTRY DATE: Entered STN: 6 Jan 1999

Last Updated on STN: 25 Jan 2002

Entered Medline: 3 Nov 1998

AΒ BACKGROUND: Present knowledge of mechanisms involved in human fertilization has uncovered a new group of pathologic conditions that have been generically named fertilization abnormalities. AIM: To determine the contribution of chromosomal alterations to in vitro fertilization failures. MATERIALS AND METHODS: A cytogenetic analysis of oocytes that were not fertilized after insemination with normal spermatozoa. Oocytes were obtained from patients subjected to in vitro fertilization in a public hospital of Metropolitan Santiago. Ovulation was induced in these patients administering GnRh-a, FSH, HMG and HCG. The double fixation technique described by Wramsby was used to obtain chromosomes. RESULTS: One hundred and seven oocytes coming from 45 women aged 25 to 42 years old were studied. The frequency of aneuploidy in these oocytes was 37.3%, with a 11.8% of hypohaploidy, a 21.6% of hyperhaploidy and a 3.9% of diploid oocytes. In hyperhaploid as well as in hypohaploid oocytes, the chromosomes involved in aneuploidy pertained to groups D. and G. CONCLUSIONS: Although the total frequency of aneuploidy is within normal ranges, the frequency of hyperhaploidy is superior to previous reports. An explanation for this finding could be that the occurrence of a lack of disjunction with chromosomal retention in the parental cell occurs with a higher frequency than that in which the chromosomes are retained in the polocyte. We also suggest that oocyte chromosomal aneuploidy could contribute to the failure of in vitro fertilization procedures.

ANSWER 8 OF 17 MEDLINE on STN ACCESSION NUMBER: 1998097568 MEDLINE DOCUMENT NUMBER: PubMed ID: 9435442

High incidence of sperm sex chromosomes TITLE:

aneuploidies in two patients with Klinefelter's

syndrome.

AUTHOR: Foresta C; Galeazzi C; Bettella A; Stella M; Scandellari C Third Chair of Medical Pathology, University of Padova, CORPORATE SOURCE:

Italy.. forestac@protec.it

SOURCE: The Journal of clinical endocrinology and metabolism,

> (1998 Jan) Vol. 83, No. 1, pp. 203-5. Journal code: 0375362. ISSN: 0021-972X.

PUB. COUNTRY: United States DOCUMENT TYPE: (CASE REPORTS)

Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals

ENTRY MONTH: 199802 ENTRY DATE: Entered STN: 24 Feb 1998

Last Updated on STN: 24 Feb 1998

Entered Medline: 9 Feb 1998

In this study we have investigated the arrangement of sex chromosomes in AΒ sperm from two severe oligozoospermic patients, apparently affected by the classic form of Klinefelter's syndrome (KS). Multicolor fluorescence in situ hybridization has been used to recognize chromosomes X, Y, and 8 in sperm from patients and 10 fertile men with normal 46,XY karyotype. patients affected by KS, we detected important numerical sex chromosome abnormalities (approximately 20%). In all normal fertile men, X- and Y-bearing spermatozoa were present in a 1:1 ratio. On the contrary, in our patients the frequency of 23,Y-bearing sperm was strongly reduced compared with that of both 23,Y sperm in the controls and 23,X sperm in the same subject affected by KS, resulting in a 23, X-/23, Y-bearing sperm ratio of 2:1. Moreover, the frequency of 24,XY disomic sperm was significantly higher in the absence of the 22,0 hypoaploidy expected from a common origin from a nondysjunction during the first meiosis in a normal 46,XY cell. In conclusion, the results of the present study demonstrate a peculiar distribution of sex chromosomes in sperm from two patients with KS, in agreement with the hypothesis that 47,XXY germ cells are able to complete the meiotic process by producing mature spermatozoa.

L5 ANSWER 9 OF 17 MEDLINE on STN ACCESSION NUMBER: 1996160505 MEDLINE DOCUMENT NUMBER: PubMed ID: 8567781

TITLE: Morphological and cytogenetic observations of unfertilized

human oocytes and abnormal embryos obtained after ovarian

stimulation with pure follicle

stimulating hormone following pituitary

desensitization.

AUTHOR: Wojcik C; Guerin J F; Pinatel M C; Bied V; Boulieu D; Czyba

J C

CORPORATE SOURCE: Department of Histology and Embryology, Warsaw Medical

Academy, Poland.

SOURCE: Human reproduction (Oxford, England), (1995 Oct)

Vol. 10, No. 10, pp. 2617-22.

Journal code: 8701199. ISSN: 0268-1161.

PUB. COUNTRY: ENGLAND: United Kingdom

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 199603

ENTRY DATE: Entered STN: 15 Mar 1996

Last Updated on STN: 15 Mar 1996

Entered Medline: 1 Mar 1996

Morphological and cytological observations of 189 unfertilized oocytes and AB 40 abnormal embryos obtained from 32 patients in a routine in-vitro fertilization programme were performed. Both the oocytes and the embryos were mounted whole to preserve the original topology of all the structural elements. With the applied protocol of ovarian stimulation associating pituitary desensitization and follicle stimulating hormone stimulation, a high degree of immaturity of the unfertilized eggs was observed in comparison with previous reports. This immaturity was deduced from the higher incidence of unfertilized eggs arrested at the germinal vesicle or metaphase I stage, as well as metaphase II oocytes with multiple metaphase plates. Nine triploid and four tetraploid embryos were analysed: except for one tetraploid embryo, all the polyploid embryos cleaved. The percentages of mononucleated blastomeres in these polyploid embryos were 57 and 27% respectively. also analysed 21 cleaving diploid embryos which exhibited a high degree of fragmentation. No more than 40% of the blastomeres contained a single nucleus. Moreover, in only one of the 21 diploid embryos could all the

blastomeres be considered normal.

L5 ANSWER 10 OF 17 MEDLINE on STN ACCESSION NUMBER: 1989008775 MEDLINE DOCUMENT NUMBER: PubMed ID: 3139703

TITLE: Chromosome anomalies in human oocytes failing to fertilize

after insemination in vitro.

AUTHOR: Bongso A; Chye N S; Ratnam S; Sathananthan H; Wong P C CORPORATE SOURCE: Department of Obstetrics and Gynaecology, National

University of Singapore.

SOURCE: Human reproduction (Oxford, England), (1988 Jul)

Vol. 3, No. 5, pp. 645-9.

Journal code: 8701199. ISSN: 0268-1161.

PUB. COUNTRY: ENGLAND: United Kingdom

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 198811

ENTRY DATE: Entered STN: 8 Mar 1990

Last Updated on STN: 8 Mar 1990 Entered Medline: 3 Nov 1988

AΒ Three-hundred-and-two unfertilized oocytes left over from successful in-vitro fertilization (IVF) attempts in 143 women (27-42 years) on a follicular stimulating hormone-human menopausal gonadotrophin (FSH -HMG) stimulation regime were subjected to chromosome analysis. Ten oocytes were degenerated with no visible chromosomes and 41 metaphases had chromosomes that were clumped together which could not be interpreted either numerically or structurally. Of the remaining oocytes, 76.6% (192/251) had a normal haploid complement (n = 23), 13% (33/251) were hypohaploid (n = 19-22), 8% (20/251) were hyperhaploid (n = 24-26), 2% (5/251) were diploid (2n = 46) and 0.4% (1/251) had structural rearrangements. The 21% aneuploidy was from 24 different patients and hypohaploid sets had chromosomes missing mainly from the A, B, C, D and G groups while the hyperhaploid sets had extra chromosomes from A, B, D, G and E groups of the human karyotype. The mean age of patients showing aneuploid oocytes was 36.7 years which was above the mean for the entire group. The aneuploidy may have been brought about by errors in oogenesis (anaphase lagging or non-disjunction) and may offer one explanation for fertilization failure and overall low pregnancy rates after IVF.

L5 ANSWER 11 OF 17 MEDLINE on STN ACCESSION NUMBER: 1987024385 MEDLINE DOCUMENT NUMBER: PubMed ID: 3490207

TITLE: Klinefelter's syndrome, mosaic

46, XX/46, XY/47, XXY/48, XXXY/48, XXYY: a case report.

AUTHOR: Al-Awadi S A; Teebi A S; Krishna Murthy D S; Othman G;

Sundareshan T S

SOURCE: Annales de genetique, (1986) Vol. 29, No. 2, pp.

119-21.

Journal code: 0370562. ISSN: 0003-3995.

PUB. COUNTRY: France

DOCUMENT TYPE: (CASE REPORTS)

Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 198611

ENTRY DATE: Entered STN: 2 Mar 1990

Last Updated on STN: 2 Mar 1990 Entered Medline: 7 Nov 1986

AB A 35-year-old male was investigated for primary

infertility. Clinical examination showed an intelligent man with

normal facial appearance and moustache and small firm testes. Testicular histopathology revealed marked atrophy of the testes with no spermatogenesis and absence of germ cells. Hormonal profile showed elevated levels of FSH,LH and low levels of testosterone. Chromosome analysis from whole blood culture showed cells with 46,XX/46,XY/47,XXY/48,XXXY/48,XXYY mosaicism. The predominant cell line was 47,XXY (87.86%). 46,XY/47,XXY mosaicism is not uncommon. However, mosaicism of multiple sex chromosome aneuploidy is rarely observed. This is the first report of mosaicism in Klinefelter's syndrome variant with five cell lines.

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ACCESSION NUMBER: 1987:273990 BIOSIS

DOCUMENT NUMBER: PREV198784015029; BA84:15029

TITLE: BRIEF CLINICAL REPORT TRISOMY XQ IN A MALE THE

ISOCHROMOSOME X KLINEFELTER SYNDROME.

AUTHOR(S): DONLAN M A [Reprint author]; DOLAN C R; METCALF M J;

BRADLEY C M; SALK D

CORPORATE SOURCE: INLAND EMPIRE GENETICS COUNSELING SERVICE, WEST 800 FIFTH

AVE, PO BOX 248, SPOKANE, WASH 99210-0248, USA

SOURCE: American Journal of Medical Genetics, (1987) Vol.

27, No. 1, pp. 189-194.

ISSN: 0148-7299.

DOCUMENT TYPE: Article FILE SEGMENT: BA LANGUAGE: ENGLISH

ENTRY DATE: Entered STN: 19 Jun 1987

Last Updated on STN: 19 Jun 1987

AB We report on a male with trisomy Xq resulting from an isochromosome Xq which is preferentially inactivated: 47, XY, + i(Xq). Six previous cases have been reported. These patients are similar to patients with classical Klinefelter syndrome (47, XXY) in that they have infertility, decreased masculinization, gynecomastia, and elevated luteinizing hormone (LH) and follide stimulating hormone (FSH) levels. They may differ in having average intelligence and normal to short stature. These findings indicate that extra copies of the long arm of X have phenotypic expression, even though activated only in early development.

L5 ANSWER 13 OF 17 EMBASE COPYRIGHT (c) 2009 Elsevier B.V. All rights reserved on STN

ACCESSION NUMBER: 2003433662 EMBASE

TITLE: Genetic analysis of sperm and implications of severe

male infertility - A review.

AUTHOR: Egozcue, Josep (correspondence); Blanco, J.; Anton, E.;

Egozcue, S.; Sarrate, Z.; Vidal, F.

CORPORATE SOURCE: Department of Cell Biology, Universitat Autonoma de

Barcelona, 08193 Bellaterra, Spain. josep.egozcue@uab.es

SOURCE: Placenta, (Oct 2003) Vol. 24, No. SUPPL. B, pp. S62-S65.

Refs: 61

ISSN: 0143-4004 CODEN: PLACDF

COUNTRY: United Kingdom

DOCUMENT TYPE: Journal; General Review; (Review)

FILE SEGMENT: 021 Developmental Biology and Teratology

028 Urology and Nephrology

LANGUAGE: English SUMMARY LANGUAGE: English

ENTRY DATE: Entered STN: 13 Nov 2003

Last Updated on STN: 13 Nov 2003

AB The use of fluorescence in situ hybridization (FISH) on decondensed sperm heads has allowed to analyse the chromosome constitution of spermatozoa in

different populations. In controls, the mean incidence of disomy (including all chromosomes) is about 6.7 per cent; diploidy increases with age, and some individuals may show a special tendency to nondisjunction. Carriers of numerical sex chromosome anomalies show a low incidence of sex chromosome disomies (2.54-7.69 per cent), and the need to screen ICSI candidates for these conditions has to be reconsidered. Carriers of inversions produce from 0 to 54.3 per cent abnormal sperm. Carriers of Robertsonian translocations produce from 3.4 to 36.0 per cent abnormal sperm, and carriers of reciprocal translocations produce from 47.5 to 81.0 per cent abnormal spermatozoa. However, carriers of translocations usually produce more abnormal embryos than expected from these figures. This may be partly related to interchromosomal effects induced by some structural reorganizations. Males with oligoasthenozoospermia, low motility and/or high FSH concentrations show frequent synaptic anomalies, resulting in the production of aneuploid and diploid sperm. Testicular sperm show extremely high rates of chromosomal abnormalities. The risk of recurrent abortion is increased by the presence of chromosome abnormalities in sperm. .COPYRGT. 2003 Elsevier Ltd. All rights reserved.

L5 ANSWER 14 OF 17 EMBASE COPYRIGHT (c) 2009 Elsevier B.V. All rights

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ACCESSION NUMBER: 2002263405 EMBASE

TITLE: Sperm aneuploidy rates in younger and older men.
AUTHOR: Luetjens, C.M.; Rolf, C.; Gassner, P.; Werny, J.E.;

Nieschlag, E. (correspondence)

CORPORATE SOURCE: Institute of Reproductive Medicine, Westphalian

Wilhelms-University, Domagkstr. 11, D-48149 Muenster,

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SOURCE: Human Reproduction, (2002) Vol. 17, No. 7, pp. 1826-1832.

Refs: 38

ISSN: 0268-1161 CODEN: HUREEE

COUNTRY: United Kingdom DOCUMENT TYPE: Journal; Article

FILE SEGMENT: 010 Obstetrics and Gynecology

022 Human Genetics

028 Urology and Nephrology

029 Clinical and Experimental Biochemistry

LANGUAGE: English SUMMARY LANGUAGE: English

ENTRY DATE: Entered STN: 8 Aug 2002

Last Updated on STN: 8 Aug 2002

Background: In order to assess the possible risk of chromosomal abnormalities in offspring from older fathers, we investigated the effects of age on the frequency of chromosomal aneuploidy rates of human sperm. Methods and results: Semen samples were collected from 15 men aged <30 years (24.8 \pm 2.4 years) and from eight men aged >60 years (65.3 ± 3.9 years) from the general population. No significant differences in ejaculate volume, sperm concentration and sperm morphology were found, whereas sperm motility was significantly lower in older men (P = 0.002). For the hormone values, only FSH was significantly elevated in the older men (P = 0.004). Multicolour fluorescence in-situ hybridization was used to determine the aneuploidy frequencies of two autosomes (9 and 18); and of both sex chromosomes using directly labelled satellite DNA probes on decondensed sperm nuclei. A minimum of 8000 sperm per donor and >330 000 sperm in total were evaluated. The disomy rates per analysed chromosomes were 0.1-2.3% in younger men and 0.1-1.8% in older men. The aneuploidy rate determined for both sex chromosomes and for the autosomes 9 and 18 were not significantly different between the age groups. Conclusions: The results suggest that men of advanced age still wanting to become fathers do not have a significantly higher risk of procreating offspring with chromosomal

abnormalities compared with younger men.

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ACCESSION NUMBER: 2001332954 EMBASE

TITLE: Studies on sperm chromosomes in patients with severe

male factor infertility undergoing

assisted reproductive technology treatment.

AUTHOR: Levron, Jacob (correspondence); Dor, Jehoshua

CORPORATE SOURCE: IVF Unit, Department of Obstetrics and Gynecology, The Chaim Sheba Medical Center, Tel Hashomer 52621, Israel.

AUTHOR: Madgar, Igal; Raviv, Gil

CORPORATE SOURCE: Male Infertility Unit, Sackler School of Medicine, Tel Aviv

University, Tel Aviv, Israel.

AUTHOR: Aviram-Goldring, Ayala; Barkai, Gad

CORPORATE SOURCE: The Center for Human Genetics, Sackler School of Medicine,

Tel Aviv University, Tel Aviv, Israel.

AUTHOR: Levron, Jacob (correspondence)

CORPORATE SOURCE: IVF Unit, Department Obstetrics and Gynecology, Chaim Sheba

Medical Center, Tel Hashomer 52621, Israel.

SOURCE: Molecular and Cellular Endocrinology, (22 Oct 2001) Vol.

183, No. SUPPL. 1, pp. S23-S28.

Refs: 33

ISSN: 0303-7207 CODEN: MCEND6

PUBLISHER IDENT.: S 0303-7207(01)00568-8

COUNTRY: Ireland

DOCUMENT TYPE: Journal; Article

FILE SEGMENT: 010 Obstetrics and Gynecology

022 Human Genetics

028 Urology and Nephrology

003 Endocrinology

037 Drug Literature Index

LANGUAGE: English SUMMARY LANGUAGE: English

ENTRY DATE: Entered STN: 11 Oct 2001

Last Updated on STN: 11 Oct 2001

AΒ The aim of the study was to determine the rate of chromosome abnormalities in testicular sperm after intracytoplasmic sperm injection due to severe male factor infertility. The study groups included patient with non-obstructive azoospermia (n=9), obstructive azoospermia (n=10), Klinefelter's syndrome (n=5) and normal controls (n=6, groups I-VI, respectively). The mean serum levels of FSH 17.5±8.2 (P<0.05), 3.5 ± 2.6 , 29.8 ± 13.0 (P<0.05) and 3.1 ± 0.4 mIU/ml, respectively. The rates of chromosome abnormalities were 19.6% (P<0.001), 8.2% (P<0.001), 6.3 and 1.6%, respectively. Chromosomes X and Y were significantly more involved in the aneuploidy than chromosome 18 in groups I and II. The present findings demonstrate a linkage between gonadal failure (high serum FSH levels) and sperm chromosome abnormalities. Our findings may explain the increased incidence of perinatal sex chromosome abnormalities found in severe male factor patients. Patients with non-mosaic Klinefelter's syndrome have comparable risk for sex chromosomes aneuploidy as the rest of the patients with azoospermia. Therefore, genetic screening during pregnancy or before embryo replacement should be carefully considered in severe male factor patient following in vitro fertilization (IVF). Copyright .COPYRGT. 2001 Elsevier Science Ireland Ltd.

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ACCESSION NUMBER: 2001016729 EMBASE

TITLE: Treating infertility in women of advanced

reproductive age.

Ascher-Walsh, C.; Klein, J.; Sauer, M.V., Dr. AIITHOR .

(correspondence)

Columbia Presbyterian Medical Center, Department of CORPORATE SOURCE:

Obstetrics/Gynecology, 622 West 168th Street, New York, NY

10032, United States.

SOURCE: Reproductive Technologies, (2000) Vol. 10, No. 4, pp.

184-188. Refs: 35

ISSN: 1528-4840 CODEN: RTEEBF

Canada

DOCUMENT TYPE: Journal; Article

FILE SEGMENT: Obstetrics and Gynecology 010

> 003 Endocrinology

LANGUAGE: English SUMMARY LANGUAGE: English

ENTRY DATE: Entered STN: 25 Jan 2001

Last Updated on STN: 25 Jan 2001

AΒ Background: Infertility is common in women who delay child-bearing. This report describes the current evaluation and treatment of infertility in women of advanced reproductive age (more than 35 years old) and discusses novel treatment strategies under current

investigation. Methods: A review of the literature indicates that the age-related decrease in female fertility is primarily a result of diminished oocyte quantity and quality. Results: Initial evaluation

should include an assessment of ovarian reserve (serum FSH

/estradiol measured at menstrual days two to three), in addition to

standard surveillance for male, uterine, and tubal or pelvic

factors. Conventional assisted reproduction outcomes are generally poor, with delivery rates less than 10% per cycle for women over 40 years old. Conclusions: Efforts at improving outcomes remain experimental and include assisted hatching of preimplanted embryos, preimplantation diagnosis of aneuploid embryos by fluorescence in situ hybridization (FISH) prior to embryo transfer, cytoplasmic or germinal vesicle transplantation using donated oocytes, and blastocyst culture. Oocyte donation has been used extensively to bypass the deleterious effects of gamete aging and remains the standard of care, with success rates ranging from 35 to 50% per cycle.

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ACCESSION NUMBER: 1998226241 EMBASE

TITLE: Intracytoplasmic sperm injection: Results from Norfolk,

USA.

AUTHOR: Oehninger, Sergio (correspondence)

CORPORATE SOURCE: Dept. of Obstetrics and Gynecology, Eastern Virginia Medical School, Norfolk, VA 23507, United States.

AUTHOR: Oehninger, Sergio (correspondence)

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> Norfolk, VA 23507, United States. Oehninger, Sergio (correspondence)

CORPORATE SOURCE: Jones Inst. Reproductive Medicine, 601 Colley Avenue,

Norfolk, VA 23507, United States.

Human Reproduction, (Sep 1996) Vol. 11, No. SUPPL. 1, pp. SOURCE:

> 73-75. Refs: 7

ISSN: 0268-1161 CODEN: HUREEE

COUNTRY: United Kingdom

DOCUMENT TYPE: Journal; Conference Article; (Conference paper)

FILE SEGMENT: 010 Obstetrics and Gynecology 028 Urology and Nephrology

037 Drug Literature Index

English LANGUAGE: English SUMMARY LANGUAGE:

AUTHOR:

ENTRY DATE: Entered STN: 27 Jul 1998
Last Updated on STN: 27 Jul 1998

AB The results of 92 consecutive couples who underwent 102 cycles of in-vitro fertilization (IVF) augmented with intracytoplasmic sperm injection (ICSI) were analysed. Inclusion criteria were previous total failed fertilization or unsuitable sperm parameters for conventional IVF. The rate of diploid fertilization was 60.9%; the implantation rate per embryo was 12.1%, and the ongoing pregnancy rate per transfer was 26.8%. None of the sperm parameters of the original or processed semen sample were correlated with ICSI outcome. Conversely, female age and basal serum concentrations of follicle stimulating hormone (FSH) had a significant impact on implantation and pregnancy rates. ICSI has become a very successful therapy in overcoming different types of male infertility.

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ALL L $^{\#}$ QUERIES AND ANSWER SETS ARE DELETED AT LOGOFF LOGOFF? (Y)/N/HOLD:y

(FILE 'HOME' ENTERED AT 10:19:24 ON 22 OCT 2009)

FILE 'MEDLINE, BIOSIS, CAPLUS, EMBASE' ENTERED AT 10:19:44 ON 22 OCT 2009 213 SEA FILE=MFE SPE=ON ABB=ON PLU=ON (FSH OR FOLLICLE(W) L1 STIMULATING(W) HORMONE) AND (XX(W) DISOMY OR YY(W) DISOMY OR ANEUPLOIDY OR DIPLOIDY) 74 SEA FILE=MFE SPE=ON ABB=ON PLU=ON L1 AND (INFERTIL?) L2 L3 43 DUP REM L2 (31 DUPLICATES REMOVED) L*** DEL 24 S L1 AND (INFERTIL?) L*** DEL L*** DEL 14 S L1 AND (INFERTIL?) L*** DEL 7 S L1 AND (INFERTIL?) L*** DEL 29 S L1 AND (INFERTIL?) 28 SEA FILE=MFE SPE=ON ABB=ON PLU=ON L3 AND (MALE) T.4 17 SEA FILE=MFE SPE=ON ABB=ON PLU=ON L4 AND PY<2004 L5 DIS IBIB ABS L5 1-17 COST IN U.S. DOLLARS SINCE FILE TOTAL ENTRY SESSION FULL ESTIMATED COST 78.11 78.33

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